Table 1

Autosomal dominant cancer syndromes

Cancer syndrome	Cutaneous manifestations	Associated features	Genetic defect
Birt-Hogg-Dubé syndrome	Fibrofolliculomas, trichodiscomas, acrochordons (skin tags), papules on buccal mucosa, collagenomas, facial angiofibroma	Renal tumours, including renal cell carcinomas and unusual histological types (can be bilateral), pulmonary cysts, pneumothorax, melanoma, parathyroid adenoma	Mutation in FLCN gene on Ch17p11.2 which codes for protein folliculin (tumour suppressor gene)
Muir-Torre syndrome (variant of hereditary nonpolyposis colorectal cancer syndrome)	Sebaceous adenoma, sebaceoma, sebaceous carcinoma, keratoacanthoma, squamous cell carcinoma, follicular cysts	Colorectal cancer, other gastrointestinal cancer, genitourinary cancer	Microsatellite instability leading to loss of MLH1 and MSH2 (DNA mismatch repair proteins)
Neurofibromatosis 1	Cutaneous and subcutaneous neurofibromas, café au lait spots, axillary freckling	Lisch nodules, scoliosis, plexiform neurofibromas, optic gliomas, phaeochromocytomas, gastrointestinal tumours, malignant peripheral nerve sheath tumours, brain tumours (meningiomas), childhood leukaemia	Mutation in NF1 gene on Ch17q11.2 which codes for neurofibromin (tumour suppressor)
Cowden syndrome (multiple hamartoma syndrome)	Trichilemmomas (well defined smooth papule/verrucoid growth), mucosal papillomatosis, acral keratosis, lipomas, haemangiomas	Breast cancer, fibrocystic breast disease, follicular thyroid cancer, endometrial cancer, uterine leiomyomas, gastrointestinal polyps	Germline intragenic mutations in PTEN gene on chromosome 10q23
Carney complex	Lentigines, blue naevus	Cardiac myxomas, testicular tumours, pancreatic cancer	Mutations in PRKAR1A gene (tumour suppressor gene)
Peutz-Jeghers syndrome	Peri-oral and oral lentigines	Hamartomatous gastrointestinal polyps, gastrointestinal cancer, pancreatic cancer	Mutation in STK11 (tumour suppressor gene)
Tuberous sclerosis	Periungual fibromas, facial angiofibromas, Shagreen patch, ash leaf macules	Renal angiomyolipomas, subependymal nodules, giant cell astrocytomas, cardiac rhabdomyomas	Mutation in TSC1 or 2 which produce hamartin and tuberin (tumour suppressor proteins)
Dysplastic naevus syndrome	Melanocytic naevi, melanoma	Melanoma, pancreatic cancer	Mutations in CDKN2A gene on chromosome 9
Howel-Evans syndrome	Palmoplantar hyperkeratosis, leukoplakia	Oesophageal cancer	Mutation in RHBDF2 on Ch17
Gorlin syndrome (nevoid basal cell carcinoma syndrome)	Basal cell carcinomas, palmar/plantar pits	Odontogenic keratocysts, childhood medulloblastomas, ovarian fibroma, cardiac fibroma, calcification of cerebral falx	Mutation in tumour suppressor gene PTCH1 on Ch9q22.3
Reed's syndrome (hereditary leiomyomatosis and renal cell cancer syndrome)	Cutaneous leiomyomas	Renal cell carcinoma, uterine leiomyosarcoma, fibroids, bladder cancer, breast cancer	Mutation in fumarate hydratase gene, which leads to accumulation of fumarate
Gardner syndrome (variant of familial adenomatous polyposis)	Epidermoid cysts, lipomas, leiomyomas, fibromas, neurofibromas	Colonic adenomatous polyps, colorectal cancer, osteomas (especially mandible), desmoid tumours, papillary thyroid cancer, hepatoblastoma, craniopharyngioma	Germline mutations in the adenomatous polyposis coli (APC) gene